

(12) INTERNATIONAL APPLICATION PUBLISHED UNDER THE PATENT COOPERATION TREATY (PCT)

10/507145

(19) World Intellectual Property Organization  
International Bureau



(43) International Publication Date  
18 September 2003 (18.09.2003)

PCT

(10) International Publication Number  
WO 03/076659 A2

(51) International Patent Classification<sup>7</sup>: C12Q 1/68

(21) International Application Number: PCT/IT03/00134

(22) International Filing Date: 6 March 2003 (06.03.2003)

(25) Filing Language: English

(26) Publication Language: English

(30) Priority Data:  
RM2002000130 8 March 2002 (08.03.2002) IT

(71) Applicant (for all designated States except US): UNIVER-  
SITA' DEGLI STUDI DI SIENA [IT/IT]; 55, Via Banchi  
di Sotto, I-53100 Siena (IT).

(72) Inventors; and

(75) Inventors/Applicants (for US only): MELONI, Ilaria  
[IT/IT]; Policlinico Le Sotte, Viale Bracci 2, 53100 Siena  
(IT). RENIERI, Alessandra [IT/IT]; Policlinico La  
Scotte, Via Bracci 2, 53100 Siena (IT).

(74) Agents: CAPASSO, Olga et al.; De Simone & Partners  
S.p.A., Via V. Bellini, 20, I-00198 Roma (IT).

(81) Designated States (national): AE, AG, AL, AM, AT, AU,  
AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN, CO, CR, CU,  
CZ, DE, DK, DM, DZ, EC, EE, ES, FI, GB, GD, GE, GH,  
GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC,  
LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW,  
MX, MZ, NI, NO, NZ, OM, PH, PL, PT, RO, RU, SC, SD,  
SE, SG, SK, SL, TJ, TM, TN, TR, TT, TZ, UA, UG, US,  
UZ, VC, VN, YU, ZA, ZM, ZW.

(84) Designated States (regional): ARIPO patent (GH, GM,  
KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZM, ZW),  
Eurasian patent (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM),  
European patent (AT, BE, BG, CH, CY, CZ, DE, DK, EE,  
ES, FI, FR, GB, GR, HU, IE, IT, LU, MC, NL, PT, RO,  
SE, SI, SK, TR), OAPI patent (BF, BJ, CF, CG, CI, CM,  
GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG).

Published:

— without international search report and to be republished  
upon receipt of that report

For two-letter codes and other abbreviations, refer to the "Guid-  
ance Notes on Codes and Abbreviations" appearing at the begin-  
ning of each regular issue of the PCT Gazette.



WO 03/076659 A2

(54) Title: DIAGNOSTIC AND THERAPEUTIC TOOLS FOR THE X-LINKED MENTAL RETARDATION SYNDROME

(57) Abstract: A nucleic acid comprising at least one fragment of the human *FACL4* gene or *FACL4* protein or functional portions thereof for diagnostic or therapeutic purposes applied to syndromes associated with mental retardation is described. Appropriate diagnostic kits are also described.